

**WHAT IS CLAIMED IS:**

1. A method for detecting the presence in a subject of a polymorphism linked to a gene associated with familial dysautonomia, said method comprising detecting a disruptive mutation in a gene of said subject encoding the I $\kappa$ B kinase-complex-associated protein.
2. The method according to claim 1, which comprises detecting a disruptive mutation in the gene encoding the I $\kappa$ B kinase-complex-associated protein which is present on chromosome 9q31.
3. The method according to claim 2, which comprises detecting a T  $\rightarrow$  C change in position 6 of the donor splice site of intron 20 of the gene encoding the I $\kappa$ B kinase-complex-associated protein which is present on chromosome 9q31.
4. The method according to claim 2, which comprises detecting a G  $\rightarrow$  C transversion of nucleotide 2390 in exon 19 of the gene encoding the I $\kappa$ B kinase-complex-associated protein which is present on chromosome 9q31.
5. The method according to claim 3 or 4, which comprises detecting said T  $\rightarrow$  C

change and/or said G → C transversion by single-strand conformational polymorphism (SSCP) analysis.

6. The method according to claim 5, wherein said SSCP analysis is carried out on a nucleic acid sequence amplified by polymerase chain reaction (PCR).

7. The method according to claim 6, wherein said nucleic acid sequence is amplified by PCR using one or more oligonucleotide primers selected from the group consisting of:

- a) GAGAACAACAAGATTCTGC (SEQ ID NO: 6);
- b) AGTCGCAAACAGTACAATGG (SEQ ID NO: 7);
- c) GCAGTTAATGGAGAGTGGCT (SEQ ID NO: 8); and
- d) ATGCTTGGTACTTGGCTG (SEQ ID NO: 9).

8. An oligonucleotide primer selected from the group consisting of:

- a) GAGAACAACAAGATTCTGC (SEQ ID NO: 6);
- b) AGTCGCAAACAGTACAATGG (SEQ ID NO: 7);
- c) GCAGTTAATGGAGAGTGGCT (SEQ ID NO: 8); and
- d) ATGCTTGGTACTTGGCTG (SEQ ID NO: 9).